

**Fig. 1**

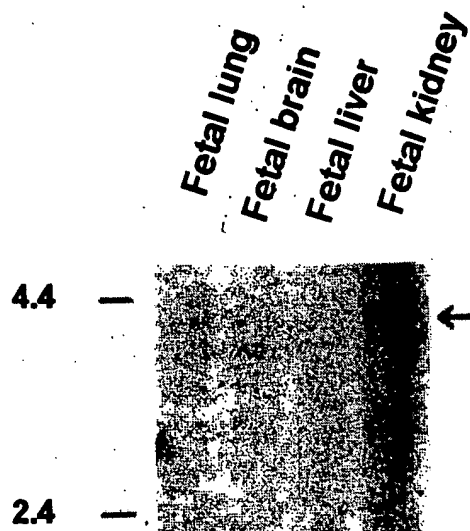


FIG. 2A

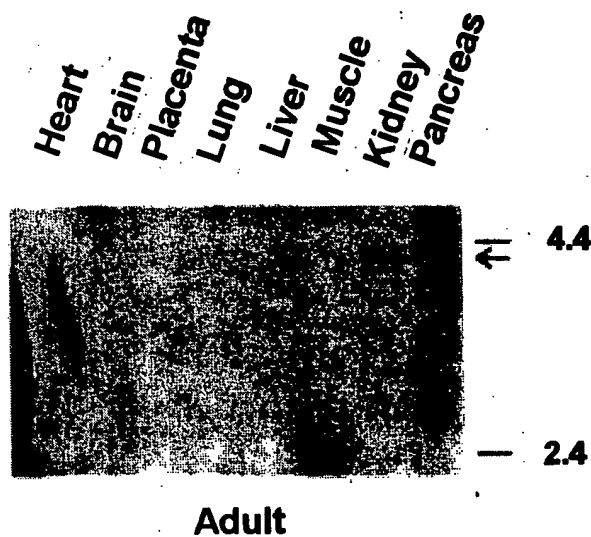
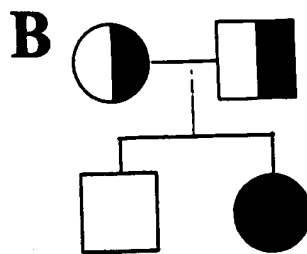
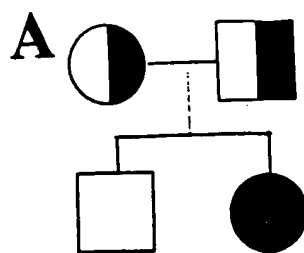


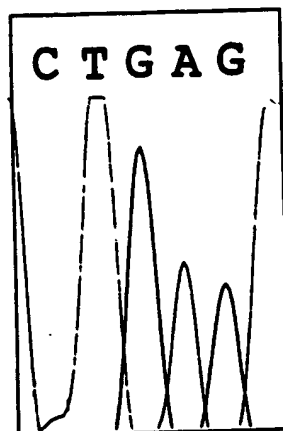
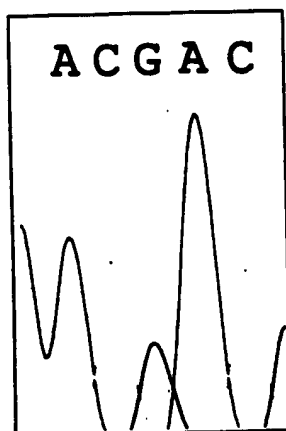
FIG. 2B

TOP SECRET

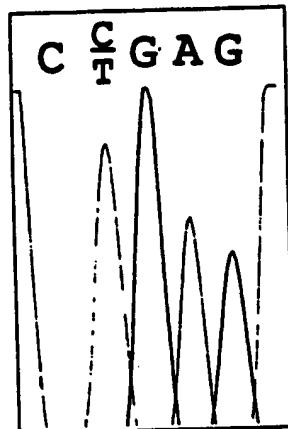
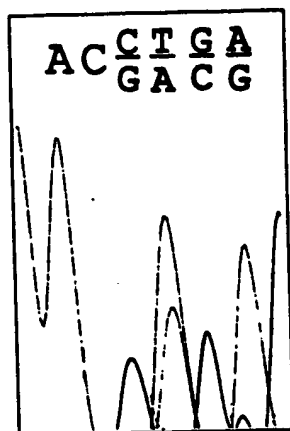
**Fig. 3**



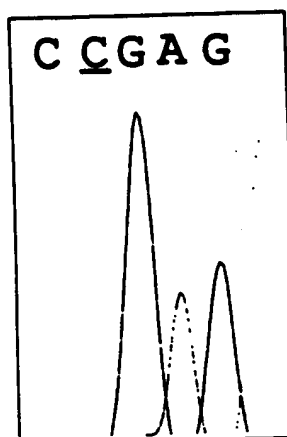
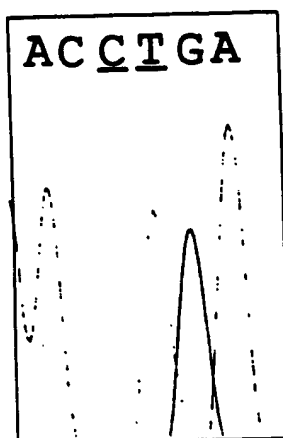
**Patient**



**Carrier parent**



**Healthy sibling**



1 MALGTTLRAS LLLGLLTEG LAQLAIPASV PRGFWALPEN LTVVEGASVE  
 51 LRCGVSTPGS AVQWAKDGLL LGPDPRIPGF PRYRLEGDPA RGEFHLHIEA  
 101 CDLSDDAEYE CQVGRSEMGP ELVSPRVILS ILVPPKLLLL TPEAGTMVTW  
 151 VAGQEVVNC VSGDAKPAPD ITILLSGQTI SDISANVNEG SQOKLFTVEA  
 201 TARVTPRSSD NRQLLVCEAS SPALEAPIKA SFTVNVLFPP GPPVIEWPGL  
 251 DEGHVRAGQS LELPCVARGG NPLATLOWLK NGQPVSTANG TEHTQAVARS  
 301 VLVMTVRPED HGAQLSCEAH NSVSAGTQEH GITLQVTFFP SAIILGSAS  
 351 QTEKNVYLS CVSKSSRPV LLRWLWGRQ LLPMEETVMD GLHGGHISMS  
 401 NLTLFARRED NGLTLTCEAF SEAFKETFK KSLILNVKYP AQLKWIEGPP  
 451 EGQKLRAQTR VRLVCLAIGG NPEPSLMWYK DSRTVTESRL PQESRRVHLG  
 501 SVEKSGSTFS RELVLVTGPS DNQAKFTCKA GQLSASTQLA VQFPPTNVTI  
 551 LANASALREG DALNLTCVSV SSNPPVNLSW DKEGERLEGV AAPRRAPFK  
 601 GSAAARSVLL QVSSRDHGQR VTCRAHSAEL RETVSSFYRL NVLYRPEFLG  
 651 EQVLVVTAVE QGEALLPVSV SANPAPEAFN HTFRGYRLSP AGGPRHRILS  
 701 SGALHLWNV T RADDGLYQLH CQNSEGTAEA RLRLDVHYAP TIRALQDPTE  
 751 VNVGGSVDIV CTVDANPILP GMFNWERLGE DEEDQSLDDM EKISRGPTGR  
 801 LRIHHAKLAQ AGAYQCIVON GVAPPARRLL RLVVRFAPQV EHPTPLTKVA  
 851 AAGDSTSSAT LHCRARGVPN IVFTWTKNGV PLDLQDPRT EHTYHOGGVH  
 901 SSSLTIANVS AAQDYALFTC TATNALGSDQ TNIQLVSISR PDPPSGLKVV  
 951 SLTPHSVGLE WKPGFDGGLP QRF CIRYEAL GTPGFHYVDV VPPQATTFTL  
 1001 TGLQPSTRYR VWLLASNALG DSGLDKGTQ LPITTPGLHQ PSGEPEDQLP  
 1051 TEPPSGPSGL PLLPVLFAIG GLLLSNASC VGGVLWQRRRL RRLAEGISEK  
 1101 TEAGSEEDRV RNEYEESQWT GERDTQSSTV STTEAEPYYR SLRDFSPQLP  
 1151 PTQEEVSYSR GFTGEDEDM FPGHLYDEVE RTYPPSGANG PLYDEVQMGF  
 1201 WDLHWPEDTY QDPRGIYDQV AGDLDTLEPD SLPFELRGHL V

FIG. 4A

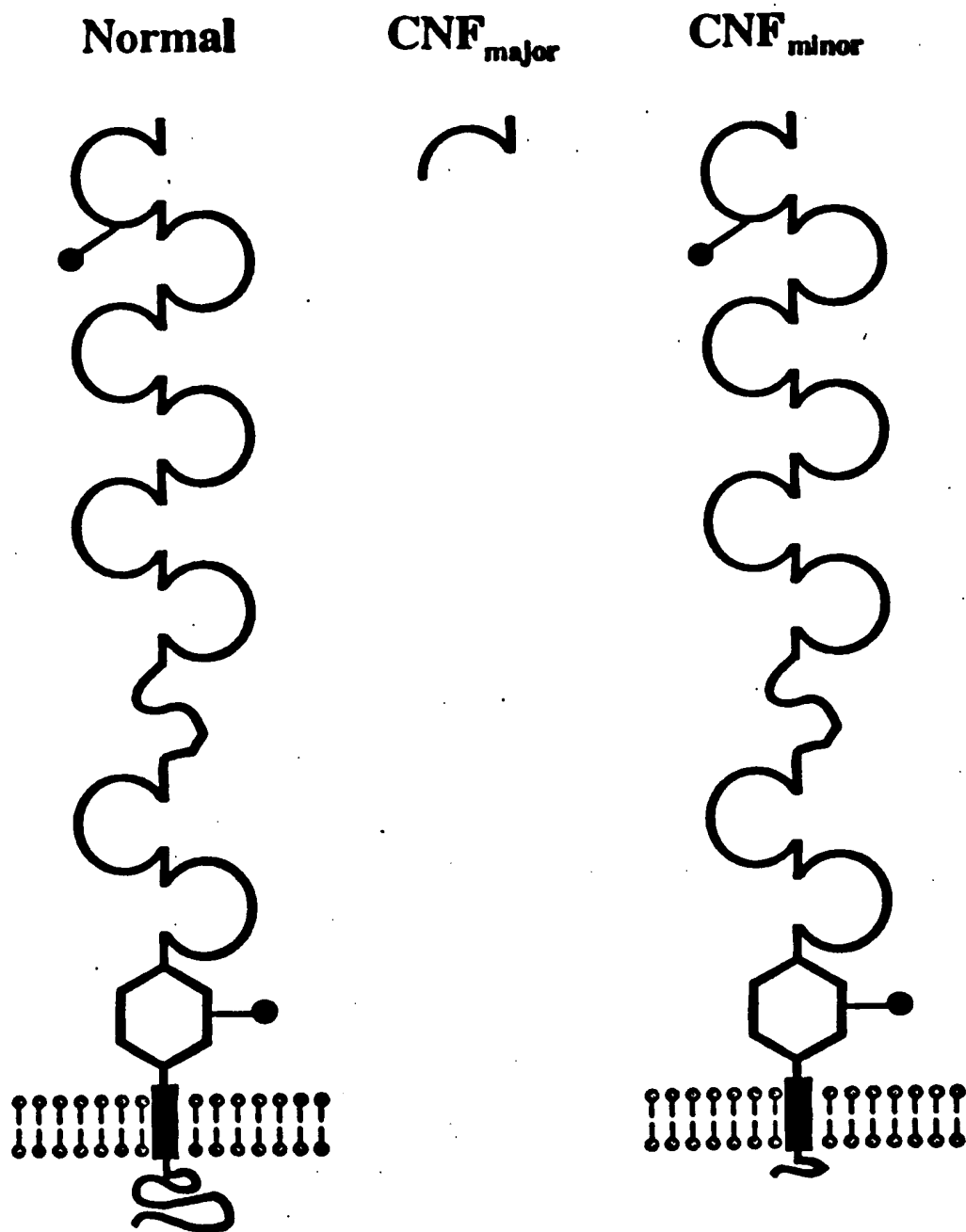
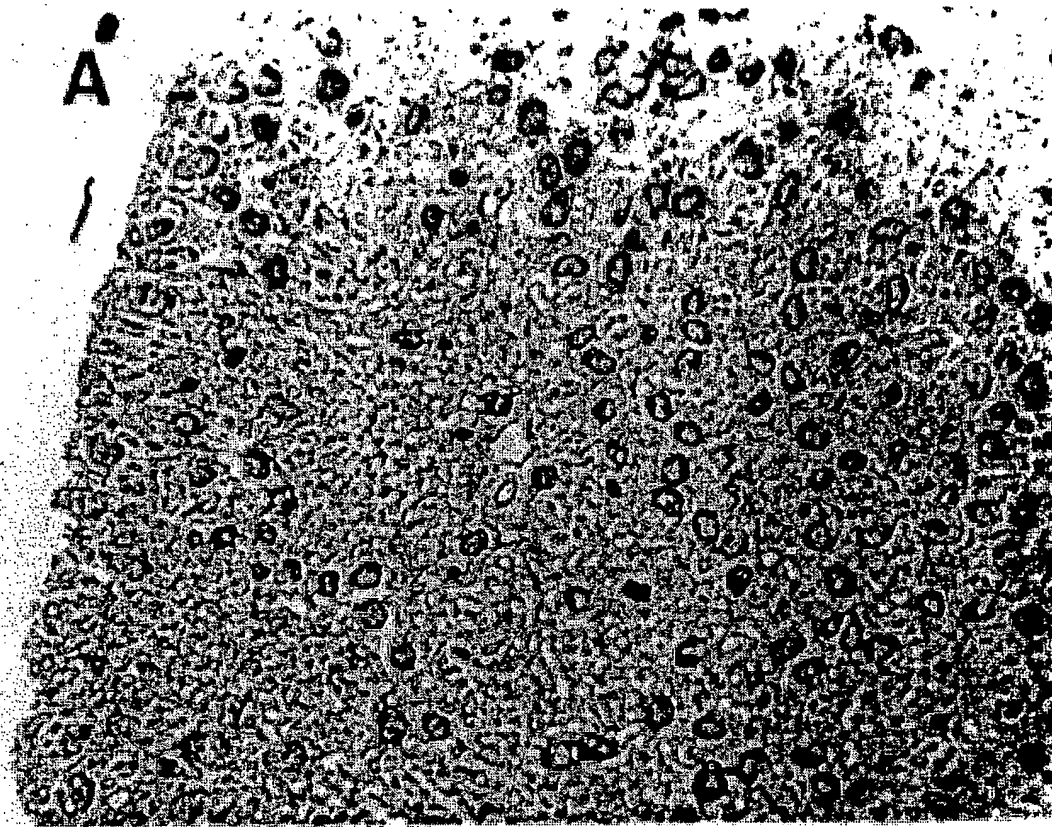
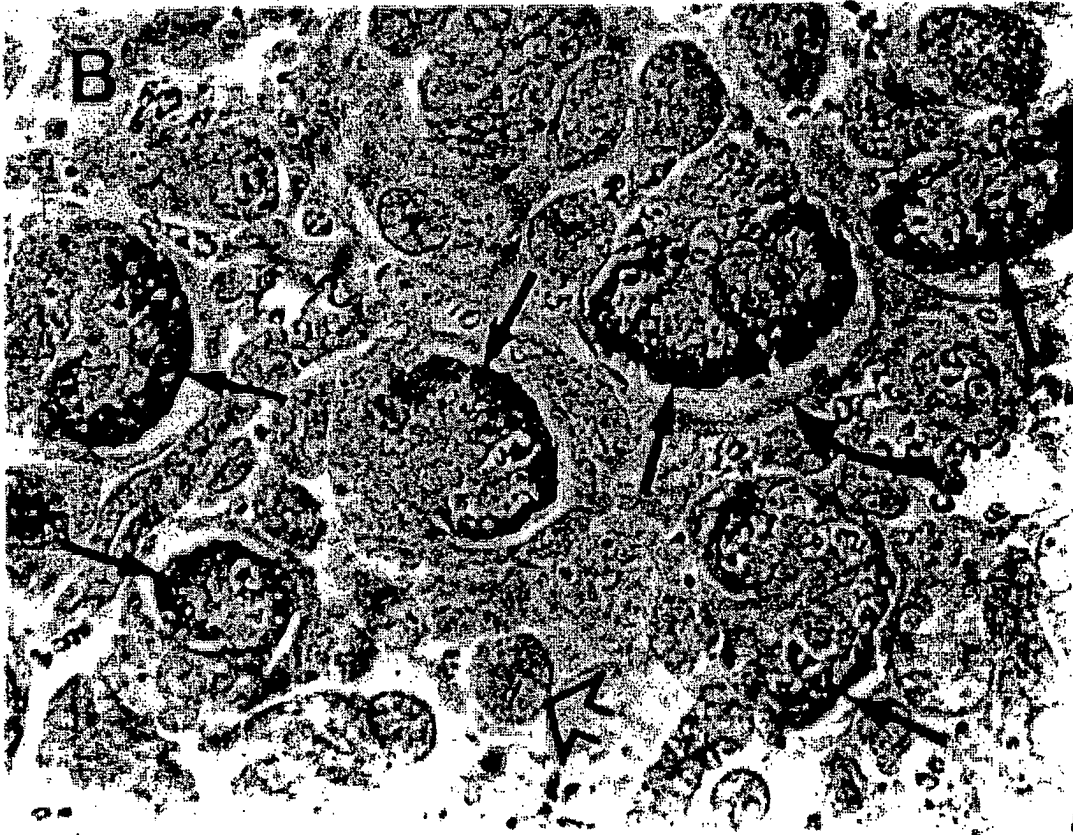


FIG. 4B



**FIG. 5A**



**FIG. 5B**